MSTN gene

myostatin

Normal Function

The *MSTN* gene provides instructions for making a protein called myostatin. This protein is part of the transforming growth factor beta (TGFβ) superfamily, which is a group of proteins that help control the growth and development of tissues throughout the body. Myostatin is found almost exclusively in muscles used for movement (skeletal muscles), where it is active both before and after birth. This protein normally restrains muscle growth, ensuring that muscles do not grow too large. Myostatin has been studied extensively in mice, cows, and other animals, and it appears to have a similar function in humans.

Researchers are studying myostatin as a potential treatment for various muscular dystrophies that cause muscle weakness and wasting (atrophy).

Health Conditions Related to Genetic Changes

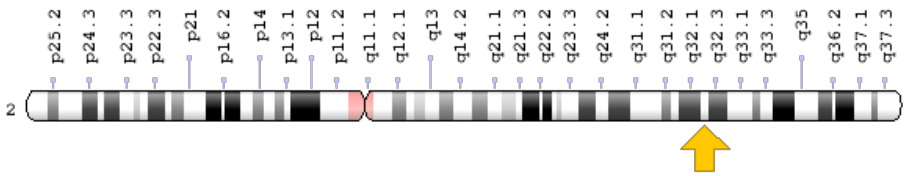
myostatin-related muscle hypertrophy

At least one mutation in the *MSTN* gene has been found to cause myostatin-related muscle hypertrophy, a rare condition characterized by increased muscle mass and strength. The mutation, which is written as IVS1+5G>A, disrupts the way the gene's instructions are used to make myostatin. As a result, cells produce little or no functional myostatin. A loss of this protein in muscle cells leads to an overgrowth of muscle tissue. It does not appear to cause any other medical problems in affected individuals.

Chromosomal Location

Cytogenetic Location: 2q32.2, which is the long (q) arm of chromosome 2 at position 32.2

Molecular Location: base pairs 190,055,700 to 190,062,729 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GDF-8
- GDF8
- GDF8_HUMAN
- growth differentiation factor 8

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Skeletal Muscle Fibers Secrete Myostatin to Limit Their Own Growth
  https://www.ncbi.nlm.nih.gov/books/NBK26853/#A4172
- Neuromuscular Disease Center, Washington University
  http://neuromuscular.wustl.edu/mother/mlarge.html#myostatinmut

GeneReviews

- Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MSTN%5BTIAB%5D%29+OR+%28myostatin%5BTIAB%5D%29%29+OR+%28%28GDF-8%5BTIAB%5D%29+OR+%28GDF8%5BTIAB%5D%29+OR+%28growth+differentiation+factor+8%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MYOSTATIN
  http://omim.org/entry/601788

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MSTN.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MSTN%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4223
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2660
- UniProt
  http://www.uniprot.org/uniprot/O14793

Sources for This Summary

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/MSTN

Reviewed: December 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services